



## neurofibromatosis type 2

Neurofibromatosis type 2 is a disorder characterized by the growth of noncancerous tumors in the nervous system. The most common tumors associated with neurofibromatosis type 2 are called vestibular schwannomas or acoustic neuromas. These growths develop along the nerve that carries information from the inner ear to the brain (the auditory nerve). Tumors that occur on other nerves are also commonly found with this condition.

The signs and symptoms of neurofibromatosis type 2 usually appear during adolescence or in a person's early twenties, although they can begin at any age. The most frequent early symptoms of vestibular schwannomas are hearing loss, ringing in the ears (tinnitus), and problems with balance. In most cases, these tumors occur in both ears by age 30. If tumors develop elsewhere in the nervous system, signs and symptoms vary according to their location. Complications of tumor growth can include changes in vision, numbness or weakness in the arms or legs, and fluid buildup in the brain. Some people with neurofibromatosis type 2 also develop clouding of the lens (cataracts) in one or both eyes, often beginning in childhood.

### Frequency

Neurofibromatosis type 2 has an estimated incidence of 1 in 33,000 people worldwide.

### Genetic Changes

Mutations in the *NF2* gene cause neurofibromatosis type 2. The *NF2* gene provides instructions for making a protein called merlin (also known as schwannomin). This protein is produced in the nervous system, particularly in Schwann cells, which surround and insulate nerve cells (neurons) in the brain and spinal cord. Merlin acts as a tumor suppressor, which means that it keeps cells from growing and dividing too rapidly or in an uncontrolled way. Although its exact function is unknown, this protein is likely also involved in controlling cell movement, cell shape, and communication between cells. Mutations in the *NF2* gene lead to the production of a nonfunctional version of the merlin protein that cannot regulate the growth and division of cells. Research suggests that the loss of merlin allows cells, especially Schwann cells, to multiply too frequently and form the tumors characteristic of neurofibromatosis type 2.

### Inheritance Pattern

Neurofibromatosis type 2 is considered to have an autosomal dominant pattern of inheritance. People with this condition are born with one mutated copy of the *NF2* gene in each cell. In about half of cases, the altered gene is inherited from an affected parent.

The remaining cases result from new mutations in the *NF2* gene and occur in people with no history of the disorder in their family.

Unlike most other autosomal dominant conditions, in which one altered copy of a gene in each cell is sufficient to cause the disorder, two copies of the *NF2* gene must be altered to trigger tumor formation in neurofibromatosis type 2. A mutation in the second copy of the *NF2* gene occurs in Schwann cells or other cells in the nervous system during a person's lifetime. Almost everyone who is born with one *NF2* mutation acquires a second mutation (known as a somatic mutation) in these cells and develops the tumors characteristic of neurofibromatosis type 2.

### Other Names for This Condition

- BANF
- bilateral acoustic neurofibromatosis
- central neurofibromatosis
- familial acoustic neuromas
- neurofibromatosis 2
- neurofibromatosis type II
- NF2
- schwannoma, acoustic, bilateral

### Diagnosis & Management

#### Genetic Testing

- Genetic Testing Registry: Neurofibromatosis, type 2  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0027832/>

#### Other Diagnosis and Management Resources

- Boston Children's Hospital  
<http://www.childrenshospital.org/conditions-and-treatments/conditions/n/neurofibromatosis-type-2>
- GeneReview: Neurofibromatosis 2  
<https://www.ncbi.nlm.nih.gov/books/NBK1201>
- MedlinePlus Encyclopedia: Acoustic Neuroma  
<https://medlineplus.gov/ency/article/000778.htm>
- MedlinePlus Encyclopedia: Neurofibromatosis 2  
<https://medlineplus.gov/ency/article/000795.htm>
- Neurofibromatosis Clinic, Massachusetts General Hospital  
<https://neurosurgery.mgh.harvard.edu/NFclinic/NF2.htm>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

### **Additional Information & Resources**

#### MedlinePlus

- Encyclopedia: Acoustic Neuroma  
<https://medlineplus.gov/ency/article/000778.htm>
- Encyclopedia: Neurofibromatosis 2  
<https://medlineplus.gov/ency/article/000795.htm>
- Health Topic: Acoustic Neuroma  
<https://medlineplus.gov/acousticneuroma.html>
- Health Topic: Neurofibromatosis  
<https://medlineplus.gov/neurofibromatosis.html>

#### Genetic and Rare Diseases Information Center

- Neurofibromatosis type 2  
<https://rarediseases.info.nih.gov/diseases/7193/neurofibromatosis-type-2>

#### Additional NIH Resources

- National Human Genome Research Institute  
<https://www.genome.gov/14514225/>
- National Institute of Neurologic Disorders and Stroke  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Neurofibromatosis-Information-Page>

### Educational Resources

- Centre for Genetics Education (Australia)  
<http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/FS62NEUROFIBROMATOSISTYPE2.pdf>
- Disease InfoSearch: Neurofibromatosis type 2  
<http://www.diseaseinfosearch.org/Neurofibromatosis+type+2/5175>
- JAMA Patient Page  
<http://jamanetwork.com/journals/jama/fullarticle/184901>
- KidsHealth from the Nemours Foundation  
<http://kidshealth.org/en/parents/nf.html>
- MalaCards: neurofibromatosis, type 2  
[http://www.malacards.org/card/neurofibromatosis\\_type\\_2](http://www.malacards.org/card/neurofibromatosis_type_2)
- My46 Trait Profile  
<https://www.my46.org/trait-document?trait=Neurofibromatosis%20type%202&type=profile>
- Orphanet: Neurofibromatosis type 2  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=637](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=637)
- The Merck Manual Home Health Handbook  
<http://www.merckmanuals.com/home/children-s-health-issues/neurologic-disorders-in-children/neurofibromatosis>
- Your Genes Your Health from Cold Spring Harbor Laboratory: Neurofibromatosis  
<http://www.ygyh.org/nf/whatisit.htm>

### Patient Support and Advocacy Resources

- Acoustic Neuroma Association  
<https://www.anausa.org/>
- Children's Tumor Foundation  
<http://www.ctf.org>
- National Organization for Rare Disorders (NORD)  
<https://rarediseases.org/rare-diseases/neurofibromatosis-2/>
- Neurofibromatosis Network  
<http://www.nfnetwork.org/>
- Resource List from the University of Kansas Medical Center  
<http://www.kumc.edu/gec/support/neurofib.html>

## GeneReviews

- Neurofibromatosis 2  
<https://www.ncbi.nlm.nih.gov/books/NBK1201>

## ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22neurofibromatosis+type+2%22+OR+%22neurofibromatosis%22>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Neurofibromatosis+2%5BMAJR%5D%29+AND+%28%28neurofibromatosis%5BTI%5D%29+AND+%282+OR+II%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

## OMIM

- NEUROFIBROMATOSIS, TYPE II  
<http://omim.org/entry/101000>

## **Sources for This Summary**

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- GeneReview: Neurofibromatosis 2  
<https://www.ncbi.nlm.nih.gov/books/NBK1201>

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